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Diaphragmatic Flutter: An Unusual Sign of Multifarious Etiologic Entities

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Abstract:

Diaphragmatic flutter (DF) is an unusual movement disorder with involuntary and repetitive contractions of the diaphragm with or without other abdominal muscle involvement. The disorder is known to occur across all ages including newborns. The etiology is diverse and so are the therapeutic options. Reaching an etiological diagnosis is considerably delayed. Response to therapy is variable and is governed by the underlying etiology. We describe three children with diaphragmatic flutter. The etiology was diverse with unrelated pathologies such as hypocalcemia, striatal necrosis, and idiopathic. All three children responded promptly and completely to calcium, high dose thiamine and biotin, and clonazepam, respectively. Our case series underscores the importance of clinical identification of such rare movement disorders. It also emphasizes that directed etiological evaluation may lead to successful amelioration of DF which is otherwise considered refractory to therapy.

Key Words:

Diaphragmatic flutter, hypocalcemia, movement disorder, myoclonus, striatal necrosis

Key Message:

Diaphragmatic flutter is an involuntary and repetitive diaphragmatic contraction. It is a rare movement disorder which often gets overlooked unless there is a high index of suspicion. Its etiology is multifarious and determines the therapeutic choices and response to therapy.

Diaphragmatic flutter (DF) is an unusual movement disorder with involuntary and repetitive contractions of the diaphragm with or without other abdominal muscle involvement. DF is variably named as diaphragmatic myoclonus, diaphragmatic tremor, respiratory myoclonus, or Leeuwenhoek disease.[1,2,10] This condition is extremely rare in the pediatric population. The etiology is multifarious. Case reports with both central as well as peripheral etiology are reported. We share our experience with three children with DF. Their clinical symptomatology, disease course, and etiology are detailed to sensitize neurologists about this entity.

Case Scenarios

Case one

A 14-year-old developmentally normal boy presented to the outpatient department with complaints of jerky movements over the abdomen noticed for the 4 four months [Video 1 and Table 1]. These movements were noticed by the caregivers as sudden jerks over mid-abdomen and were not perceived by the child himself. Subsequently, these movements were noted throughout the day, used to subside during sleep, and had no relation to exertion or stress. There were no other complaints such as seizures, abnormal movements of other body parts, hiccups, pain, or breathing difficulty. He was academically doing well with no evidence of any stress or behavioral abnormalities. Given the myoclonic nature of movements and the location over the diaphragmatic area, a clinical diagnosis of DF was made and ultrasonography (USG) with entrainability was planned. In USG abdomen, sudden jerky movements originating from the diaphragm were seen with some entrainability. Serum biochemistry and MRI of brain and spine were unremarkable. The diagnosis of DF was confirmed and the child was started on clonazepam. Etiology of DF could not be ascertained and thus was labeled as idiopathic. At 3 months follow-up, the child is doing well without any recurrence.

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Case two

A 10-year-old girl presented to the emergency department with acute onset of abdominal pain along with irregular hyperkinetic abdominal movements for the last 12 hours. It was not associated with any vomiting, diarrhea, weakness, or respiratory difficulty. She had no voluntary control over these movements. She was a typically developing girl and had no known comorbidity or any recent or prolonged drug intake. No similar events in the past have ever occurred. On examination, there was continuous movement of the abdominal wall in form of rhythmic contractions at frequency of 180–240/min [Video 2]. It persisted even while holding her breath and during talking, without interfering with the speech. She had rachitic rosary and bilateral wrist widening with positive Chovstek's and Trousseau's signs. Her serum ionized calcium was 0.23 mmol/L (normal value: 1.22–1.37 mmol/L) and the rest of the emergency investigations were within normal limits. Calcium gluconate infusion was initiated following which the abnormal movements subsided. Later workup revealed a low serum parathormone level and hypoparathyroidism was diagnosed. She is currently on oral calcium therapy and the movements never recurred.

Case three

A 9-year-old boy was referred to our hospital for evaluation of acute onset encephalopathy with new-onset jerky abdominal movements. The boy was a typically developing child who had a new-onset, refractory, right focal status epilepticus, and low-grade fever, 12 days back. In an outside medical facility, status was aborted with midazolam and valproate in about 3 hours. Four days later, the child became afebrile but developed unsteady gait and difficulty in bearing weight on lower limbs, which progressed to paraplegia over the next 12 hours. By day eight of illness, the parents noticed abnormal jerky movements of the left upper abdomen. Subsequently, the child was shifted to our center. On examination, he was conscious, oriented, and interactive. Frequent, involuntary, irregular, jerky movements of 120 to 150/min were noticed over his left upper abdomen. These jerks were independent of his regular breathing [Video 3] and decreased in frequency and severity during sleep. MRI brain with contrast revealed homogenous, symmetrical T2/FLAIR hyperintensity in bilateral caudate and putamen (bilateral striatal necrosis) without meningeal enhancement [Figure 1]. MRI thoracic and cervical spine was normal. CSF study including lactate was normal. Infective etiological evaluation for scrub typhus, HSV, HBV, HCV, HIV, Legionella, and Mycoplasma serology were unyielding. Neurometabolic possibilities such as biotin thiamine responsive basal ganglia disorder, mitochondrial cytopathy, etc., were considered at this juncture and the child was started on thiamine and biotin. Metabolic workup including serum ammonia, blood tandem mass spectrometry (TMS), and urine gas chromatography mass spectrometry (GCMS) was within normal limits. The child improved symptomatically though DF persisted at the time of discharge. On follow-up at 8 weeks, DF had resolved. Genetic confirmation of the underlying diagnosis could not be done due to financial constraints.

Discussion

Diaphragmatic flutter or diaphragmatic myoclonus most commonly presents with abnormal jerky movements of the upper abdomen. Other presentations including epigastric

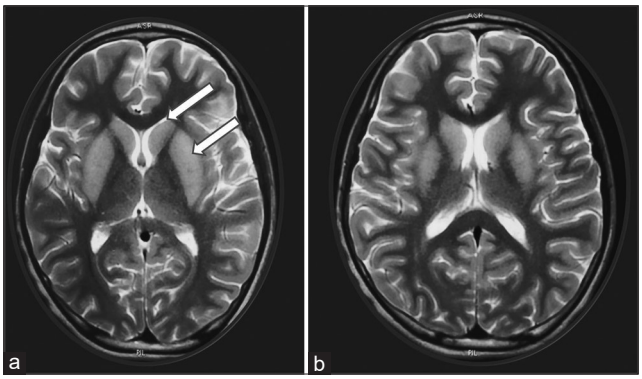

Figure 1: (a and b) MRI brain 3 Tesla of case 3: T2 weighted axial sections (a and b) showing hyperintensity in bilateral caudate head and putamen (arrows)

Table 1: Summary of clinical details, investigations, and clinical course of the 3 subjects

	Case 1	Case 2	Case 3
Age and sex	14 years/M	10 years/F	9 years/M
Presenting complaint	Abdominal jerks	Abdominal pain	Encephalopathy and focal seizures
Duration of DF*	12 days	12 hours	4 months
Frequency of DF	30-40/min	180-240/min	120-150/min
Interference with respiration or speech	No	No	No
Voluntary Suppression	No	No	No
Effect on holding of breath	Decrease in frequency	Decrease in frequency	Could not be ascertained
Gastro-esophageal symptoms	No	No	No
Effect of sleep	Subsides	-	Subsides
MRI brain	Normal	-	Bilateral striatal necrosis
Etiology	Unknown	Hypocalcemia due to hypoparathyroidism	Bilateral Striatal Necrosis possibly due to biotin thiamine responsive basal ganglia disease
Treatment	Clonazepam	Calcium supplementation	Biotin and thiamine
Course	DF subsided in one week. No recurrence on clonazepam	DF subsided within hours and no recurrence on calcium supplements	DF subsided in 4 weeks and no recurrence on biotin and thiamine supplements

*Diaphragmatic flutter

pain, palpitations, and dyspnea are also reported. Owing to the rarity, this disorder is often missed and may even take years to reach the correct diagnosis.[11,12] Nevertheless, the unique manifestation in the form of isolated jerky abdominal movements is present in only a few other conditions. Belly dancer's dyskinesia (BDD) or belly dancer syndrome is such a condition where the abdominal muscles undergo rhythmic contractions leading to undulating movements of the anterior abdominal wall, displacing the position of the umbilicus and resembles a belly dancer. But the literature shows that the terms DF and BDD are used often interchangeably obscuring a proper delineation between the two. Iliceto *et al*.[1] reported five cases of abnormal abdominal movements of which only one case was diagnosed as DF and the rest were designated as BDD. The authors opined that DF may be a variant of palatal myoclonus syndrome with dysfunction of the dentato-rubro-olivary pathway and BDD due to a more peripheral pathology. DF is considered as a palatal tremor variant with the most severe presentation with respiratory compromise designated as "symptomatic respiratory myoclonus" and the mildest type known as isolated diaphragmatic tremor.[2] Authors differ about the myoclonus generator in cases of DF and about the type of myoclonus. DF associated with encephalitis, cerebellitis, and spinal cord trauma exemplifies a central cause,[3] whereas many cases are reported following pleurisy and pericarditis suggesting a peripheral cause.[4] The investigations recommended for diagnosis and delineating the muscle of origin are fluoroscopy, EMG, or USG abdomen.[2] In our series, two subjects (case one and two) presented with only abnormal abdominal movements, whereas case three had encephalopathy and focal seizures with abdominal jerks noticed incidentally. In all cases, movements involved the upper abdomen only without the displacement of the umbilicus. In case two, the movements subsided soon following calcium infusion and so USG or fluoroscopy was not possible for confirming diaphragmatic origin. In case three, USG confirmation of the diagnosis could not be done as other major clinical signs and symptoms drew our attention.

The underlying etiology of DF in the majority of cases is unknown leading to the assumption of it being a functional movement disorder. It is strengthened by the fact that the movements are exacerbated by stress and most cases have normal neuroimaging and investigations. Etiology was multifarious in our series [Table 1]. A definite etiology could not be ascertained in case one; however, lack of any behavioral issues or stressors and a definite diaphragmatic origin on ultrasonography suggest an organic pathology. Case two had a systemic or peripheral cause in the form of hypocalcemia secondary to hypoparathyroidism. Hypocalcemia is an extremely unusual etiology of DF. Though muscle cramps, carpopedal spasms, tetany, laryngospasm, and seizures are known, there just exists a single report of hypocalcemia leading to DF in humans in English literature.[5] Another case study reported a dog with DF diagnosed to have hypocalcemia secondary to hypoparathyroidism.[6] Case three had a central etiology as the MRI brain showed bilateral striatal involvement. Interestingly, the child had unilateral (left hemidiaphragmatic) flutter which may suggest additional microstructural or functional affection of the dentato-rubro-olivary pathway, the presumed site of origin of DF. Since the encephalopathy and seizures along with DF subsided following high dose thiamine and biotin therapy, a probable diagnosis of striatal necrosis due to biotin-thiamine responsive basal ganglia disease (BTBGD) was kept, though a genetic confirmation of the same could not be done.

Being a rare entity there are no consensus guidelines for the treatment of DF. Phenytoin, carbamazepine, clonazepam, haloperidol, and aripiprazole are among the few drugs tried in individual cases with variable response. Neonatal onset DF is of particular concern as DF starts as early as day one of life and leads to respiratory distress which responds to CPAP and chlorpromazine.[7,8] Children and adults with DF who do not respond to conservative management with drugs are subjected to phrenic nerve crushing or botulinum toxin injection.[9] In our series, DF subsided in all the three cases with albeit different and unusual drugs (calcium injection, oral biotin and thiamine, and clonazepam respectively). None of the cases had a recurrence of DF at last follow-up.

Conclusion

Our series sensitizes pediatricians and child and adult neurologists about this unusual and rare movement disorder that has a long latent period before diagnosis. It is also highlighted that DF is potentially treatable and has a diverse etiology with both central and peripheral or systemic causes. Timely recognition of DF may dissuade clinicians from ordering irrelevant investigations and end their quest for a correct diagnosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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